Original Article

A Hospital Based Cross Sectional Study of Midline Cutaneous Lesions in Neonates and its Association with Spinal Dysraphism Detected Using Ultrasound

Abstract

Background: Spinal dysraphism occurs due to incomplete fusion of the midline mesenchymal, bony, or neural elements of the spine. The defects in the spinal cord can be associated with skin lesion since both have same embryonic origin. **Aims and Objectives:** This study was conducted to determine the association of midline and paramedian cutaneous lesions with spinal dysraphism by using spinal ultrasonography. **Materials and Methods:** Two thousand apparently healthy neonates were screened in the postnatal ward of a tertiary care center in South India. Those neonates with cutaneous lesions in the midline and paramedian region were screened for evidence of spinal dysraphism by using spinal ultrasonography. **Results:** Among 2000 neonates, 120 (6%) had at least one cutaneous lesion, of which 114 (5.7%) were in the midline and 6 (0.3%) were on the paramedian region of dorsal and ventral aspect of the body. Among these neonates, two cases had more than one skin lesions. The most common cutaneous lesion observed was typical dimple (82, 68%) followed by hypertrichosis (12, 10%). Ultrasonography revealed spinal anomaly in six (5%) of them. The cutaneous lesions associated with spinal dysraphism were obvious midline swelling, dermal sinus, and multiple skin lesions. **Conclusion:** Congenital midline and paramedian skin lesions may be the marker of spinal dysraphism. In the presence of such cutaneous lesions, only 5% of them had associated spinal anomaly in our study.

Keywords: *Midline skin lesions, sonography, spinal dysraphism*

M. R. Resmi, Devinder Mohan Thappa, Laxmisha Chandrashekar[1], Nishad Plakkal[2], A. Ramesh[3]

Departments of Dermatology, [1]Dermatology and STD, [2]Neonatology and [3]Radiodiagnosis, Jawaharlal Institute of Postgraduate Medical Education and Research (JIPMER), Pondicherry, India

Introduction

Spinal dysraphism (SD) refers to congenital anomalies that are caused by an incomplete fusion of the midline mesenchymal, bony, or neural elements of the spine. The defects in the spinal cord can be associated with skin lesions since both have the same embryonic origin. Occult spinal dysraphism (OSD) refers to SD in which the skin covers the neural tissue. This occurs commonly at the level of S1, S2, or both.[1]

Congenital midline and paramedian skin lesions may be a marker of SD. The clinical spectrum of OSD is broad, ranging from skin anomalies to motor, urinary, or bowel dysfunctions.[2,3] Notably however, symptoms related to OSD are often not clinically obvious at birth and are usually subsequently revealed by a radiographic or physical examination. Therefore, affected patients present with delayed neurologic, urologic, and orthopedic symptoms and may have irreversible impairment.[2,3] Hence, in the presence of such cutaneous lesions, radiologic investigations are generally recommended to detect possible occult SD. It helps in early diagnosis of SD and in preventing complications.[1]

Magnetic resonance imaging is the most sensitive radiologic investigation for detecting SD; however, ultrasonography (USG) is an excellent noninvasive alternative in infants less than 6 months of age.[4] There is paucity of data and literature on this issue. Existing literature is predominantly based on case reports and case series. Hence, we undertook this study to determine the frequency of midline cutaneous lesions and to find out which midline cutaneous lesions actually could act as marker of SD by spinal USG.

How to cite this article: Resmi MR, Thappa DM, Chandrashekar L, Plakkal N, Ramesh A. A hospital based cross sectional study of midline cutaneous lesions in neonates and its association with spinal dysraphism detected using ultrasound. Indian Dermatol Online J 2021;12:408-11.

Received: 14-Sep-2020. **Revised:** 26-Nov-2020. **Accepted:** 25-Jan-2021. **Published:** 12-May-2021.

Address for correspondence:
Dr. Devinder Mohan Thappa, Department of Dermatology, Jawaharlal Institute of Postgraduate Medical Education and Research (JIPMER), Pondicherry - 605 006, India.
E-mail: dmthappa@gmail.com

Access this article online

Website: www.idoj.in

DOI: 10.4103/idoj.IDOJ_748_20

Quick Response Code:

Materials and Methods

This was a hospital-based cross-sectional study done in neonates within 48 h of birth born in the Women and Children hospital, at a tertiary care center south India during the study period of 3 years. The institute ethics committee clearance was obtained before undertaking the study (Ethical clearance number: JIP/IEC/2015/17/663 dated 9.9.2015).

After obtaining written informed consent from either of the parents, neonates within the first 48 h of life were screened for midline and paramedian lesions over both dorsal and ventral aspect of the body including scalp by a trained dermatologist. There were no exclusion criteria. The demographic details, maternal antenatal drug history, and clinical findings pertaining to the number, distribution, and morphology of the lesions were recorded. In neonates with midline cutaneous skin lesions, USG was done using 7.5 MHz color Doppler USG scanner Vivid S5 [GE Healthcare] for evidence of SD in the Radiodiagnosis department. Those with SD were referred to neurosurgeon and orthopaedician for further evaluation.

The data collected were tabulated in Microsoft Excel Worksheet and analyzed using the IBM SPSS 20.0 statistical software and descriptive statistics were used for analysis. The association between various cutaneous findings in those with and without SD was analyzed using Fischer exact test. The statistical analysis was carried out at 5% level of significance and a value of $P < 0.05$ was considered statistically significant.

Results

Two thousand neonates with a mean age of 23.98 ± 0.67 h (ranging from 12 to 36 h) were screened for midline and paramedian cutaneous lesions. It included 1116 (55.8%) boys and 884 girls (44.2%). Of the 2000 neonates, 120 (6%) had cutaneous lesions with 114 being midline and 6 being paramedian. Two of 120 had more than one cutaneous lesions. Among the neonates with cutaneous lesions, 59 (49.2%) were boys and 61 (50.8%) were girls.

The mean age of the mothers of the recruited neonates was 24.86 ± 3.21 years. There was no significant association observed between maternal age and the presence of cutaneous lesions (Chi-square test, $P = 0.917$). Of the 120 neonates with cutaneous lesions, it was observed that 12 mothers had gestational diabetes mellitus, 7 had hypertension and one mother had antenatal bleeding per vagina. No significant association was found between maternal illness and cutaneous lesions (Chi-square test, $P > 0.05$). No significant association was found between maternal drug intake and cutaneous lesions or SD using Chi-square test with $P > 0.05$. None of the mothers during the antenatal period had a history of possible teratogenic drug intake.

Of the 120 neonates with cutaneous lesions, the most common cutaneous lesion observed was typical dimple seen among 82 neonates (68.33%). The other lesions were hypertrichosis (12, 10%), atypical dimple (7, 5.83%), café au lait macules (5, 4.17%), congenital melanocytic nevi (4, 3.3%), hemangioma (3, 2.5%), aplasia cutis (3, 2.5%), swelling (2, 1.6%), dermal sinus, acrochordon, hypopigmentation, and sebaceous nevus (1 neonate each) [Tables 1 and 2].

Among the 2 neonates with multiple lesions, one had hypertrichosis in lumbar and sacral region with hemangioma in lumbar region and the other had swelling over lumbar region with hypertrichosis in sacral region.

The prevalence of SD in 2000 neonates screened was 0.3%. Six of the 120 (5%) neonates with cutaneous lesions including the two with multiple lesions showed

Table 1: Frequency of various cutaneous lesions in the neonates

Cutaneous lesions	*n*=120 (%)	*n*=2000 (%)
Typical dimple	82 (68.33)	82 (4.1)
Hypertrichosis	12 (10)	12 (0.6)
Atypical dimple	7 (5.8)	7 (0.35)
Hyperpigmentation	5 (4.16)	5 (0.25)
Nevi	4 (3.3)	4 (0.2)
Haemangioma	3 (2.5)	3 (0.15)
Aplasia cutis	3 (2.5)	3 (0.15)
Swelling	2 (1.6)	2 (0.1)
Dermal sinus	1 (0.83)	1 (0.05)
Acrochordon	1 (0.83)	1 (0.05)
Hypopigmentation	1 (0.83)	1 (0.05)
Sebaceous nevus	1 (0.83)	1 (0.05)

Total number of lesions - 122; as 2 neonates had more than one lesions

Table 2: Site of various cutaneous lesions in the neonates

Cutaneous lesions	Site	Number of neonates (*n*=120)
Typical dimple	Sacral	82
Hypertrichosis	Sacral	6
	Lumbar	3
	Lumbo-sacral	3
Atypical dimple	Sacral	7
Hyperpigmentation	Lumbar	4
	Abdomen	1
Melanocytic nevi	Face	1
	Neck	1
	Lumbar	1
	Sacral, lumbar and abdomen	1
Haemangioma	Scalp	2
	Lumbar	1
Aplasia cutis	Scalp	3
Swelling	Lumbar	2
Acrochordon	Sacral	1
Hypopigmentation	Face	1
Sebaceous nevus	Face and scalp	1

evidence of SD on USG. None of the neonates had any neurological defects at presentation. All neonates with ultrasonographic evidence of SD were males, had midline lesions [Figures 1a, b and 2a, b]. The findings on USG in these neonates are presented in Table 3. Apart from single lesion on the scalp, rest were in lumbosacral region. The finding of obvious swelling (two cases), dermal sinus (one case), and more than two lesions (2 cases) were invariably associated with SD in our study. None of the neonates had any neurological deficit or orthopaedic (spinal and limb) deformities. No obvious urogenital malformations were found.

Discussion

SD is caused by an incomplete fusion of the midline mesenchymal, bony, or neural elements of the spine.[5] The incidence of SD is 1-3/1000 live birth.[6] SD can have associated cutaneous stigmata in 4.2%–7.2% of all neonates. In infants with cutaneous markers, association with SD has been found in 3%–8% of cases.[7]

Choi *et al*.[8] in their systematic review and meta-analysis of 15 studies involving 6558 patients, found the pooled proportion of OSD detected by ultrasound among cases with cutaneous stigmata to be 2.8%. A stronger association with OSD was found in patients with combined stigmata and atypical dimple. In the recently published study from India, a total of 1000 neonates were examined over a period of 3 years, of which cutaneous signs of SD were seen in 135 (13.5%) new-borns.[4] Sacral dimple was most seen in 128 (12.8%) neonates, followed by meningomyelocele in 5 (0.5%), dermoid cyst in 1 (0.1%) and acrochordons in 1 (0.1%) neonate. The diagnosis was made clinically, and no radiological investigation was carried out on them. In our study of 2000 neonates screened, 120 (6%) had at least one median or paramedian cutaneous lesions, comparable with the reported prevalence of 7.2%–8% from other centers.[9,10] The most common cutaneous lesion found in our study was typical sacral dimple which was present in 68.3% of neonates with cutaneous lesions followed by hypertrichosis in 10%. Kriss and Desai[9] in their study on 207 neonates also had observed typical dimple commonly (77.3%), followed by atypical dimple (6.2%) and hypertrichosis (4.8%). This finding was different when compared to the study on 144 neonates by Henrique *et al*.[10] who reported hypertrichosis (67 neonates) as a common stigma followed by the shallow dimple (32 neonates) and deep gluteal fold (30 neonates).

In our study, 6 (5%) neonates with cutaneous lesions including the two with multiple lesions showed evidence of spinal anomaly/dysraphism. The most common spinal anomaly observed on USG was splaying of posterior elements with meningocele. Finding a dermal sinus or midline swelling in lumbosacral region was invariably associated with SD in our study. None of the neonates with atypical dimple had SD while one neonate with typical dimple had an ultrasonographic finding of filar lipoma. Studies in the past have revealed a low association with typical dimple and SD.[7,11,12] Guggisberg *et al*.[13] noted that dermal sinus to be a high-risk lesion for SD (reported spinal anomaly in 5 of 6 neonates with dermal sinus).

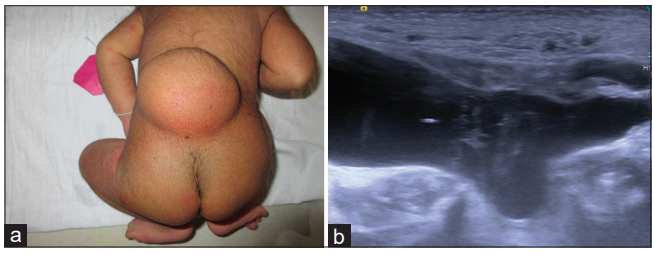

Figure 1: (a) Swelling over lumbar region with hypertrichosis in sacral region. (b) Ultrasonography of same neonate (a) showing meningocele

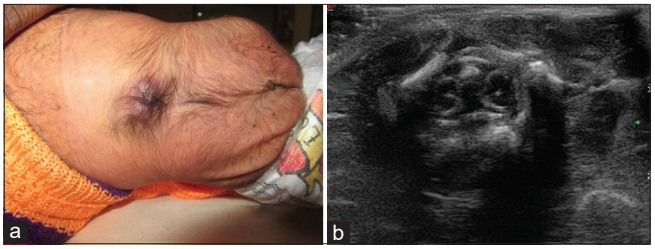

Figure 2: (a) Hypertrichosis in lumbar and sacral region with hemangioma in lumbar region in neonate. (b) Ultrasonography of same neonate (a) showing splaying of posterior elements with diastematomyelia

Table 3: Ultrasonographic findings in 6 neonates with spinal anomaly

Midline skin lesion	Ultrasonographic findings	Diagnosis
Aplasia cutis (One of three)	Lower lumbar vertebrae show defect between the lamina, no herniation, spina bifida occulta.	Aplasia cutis of scalp with spina bifida occulta.
Typical dimple (One of 82)	Filar lipoma	Typical dimple with filar lipoma
Dermal sinus (One of one)	External lesion communicating with the coccyx through a 3 mm band like structure with splaying of posterior elements	Dermal sinus with spina bifida occulta
Swelling in the back (Case 1 of two)	Splaying of posterior elements with meningocele in the lower lumbar region.	Swelling of back with meningocele
Swelling in the back (Case 2 of two) with Hypertrichosis (Case 1 of 12)	Defect in posterior elements of spine with meningocele.	Swelling of the back with meningocele
Haemangioma (One of three) with hypertrichosis (Case 2 of 12)	Splaying of posterior elements with meningocele in lower lumbar region with bony spur dividing the cord.	Hemangioma with hypertrichosis with meningocele with diastematomyelia.

We observed two of 12 cases with hypertrichosis and SD, though many previous studies[12,13] reported a high association between the two.

Occurrence of multiple cutaneous lesions as observed in 2 neonates (of 120 cases) in our study were associated with SD. Among these two neonates; a combination of hypertrichosis with swelling on the dorsal aspect of the trunk were found in one neonate, and hypertrichosis with hemangioma found in other. Both neonates had splaying of posterior elements of the spinal cord with meningocele, one in addition had diastematomyelia. This observation was recorded in lower frequency than many other studies done previously. Henrique *et al*.[10] noted 27% of neonates with multiple skin lesions whereas Kriss and Desai[9] reported multiple cutaneous lesions in 9 of 207 (4.4%) neonates. Sardana *et al*.[12] also noted that a combination of two or more skin lesions in the midline was a strong marker of SD. In their study, 10 of 24 children with multiple cutaneous lesions had SD as compared to 40 of 156 with single cutaneous lesions. We did not observe any significant association between maternal age or maternal illness with SD or cutaneous lesions in our study. But Vieira and Taucher[14] in their meta-analysis investigating the association between maternal age and neural tube defects reported an association between increased maternal age and spinal bifida.

Limitations of the study

The number of cases with SD in association with midline skin lesions were too small to draw meaningful comparison by statistical analysis. Magnetic resonance imaging was logistically not possible; however, the same could have been done at 6 months of age or later.

Conclusion

Midline or paramedian cutaneous lesions like obvious swelling, dermal sinus, and multiple skin lesions were associated with spinal anomaly/dysraphism in only 5% of the neonates by USG.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Netto JMB, Bastos AN, Figueiredo AA, Pérez LM. Spinal dysraphism: A neurosurgical review for the urologist. Rev Urol 2009;11:71-81.
2. Satar N, Bauer SB, Shefner J, Kelly MD, Darbey MM. The effects of delayed diagnosis and treatment in patients with an occult spinal dysraphism. J Urol 1995;154:754-8.
3. Gharedaghi M, Samini F, Mashhadinejad H, Khajavi M, Samini M. Orthopedic lesions in tethered cord syndrome: The importance of early diagnosis and treatment on patient outcome. Arch Bone Jt Surg 2014;2:93-97.
4. Taj FT. Cutaneous markers of spinal dysraphism: A cross sectional study. Indian J Paediatr Dermatol 2018;19:215-9.
5. Drolet B. Birthmarks to worry about. Cutaneous markers of dysraphism. Dermatol Clin 1998;16:447-53.
6. Venkataramana NK. Spinal dysraphism. J Pediatr Neurosci 2011;6:S31-40.
7. Robinson AJ, Russell S, Rimmer S. The value of ultrasonic examination of the lumbar spine in infants with specific reference to cutaneous markers of occult spinal dysraphism. Clin Radiol 2005;60:72-7.
8. Choi SJ, Yoon HM, Hwang JS, Suh CH, Jung AY, Cho YA, *et al*. Incidence of occult spinal dysraphism among infants with cutaneous stigmata and proportion managed with neurosurgery: A systematic review and meta-analysis. JAMA Netw Open 2020;3:e207221.
9. Kriss VM, Desai NS. Occult spinal dysraphism in neonates: Assessment of high-risk cutaneous stigmata on sonography. Am J Roentgenol 1998;171:1687-92.
10. Henriques JG de B, Pianetti G, Henriques KSW, Costa P, Gusmão S. Minor skin lesions as markers of occult spinal dysraphisms--prospective study. Surg Neurol 2005;63(Suppl 1):S8-12.
11. Kucera JN, Coley I, O'Hara S, Kosnik EJ, Coley BD. The simple sacral dimple: Diagnostic yield of ultrasound in neonates. Pediatr Radiol 2015;45:211-6.
12. Sardana K, Gupta R, Garg VK, Mishra D, Mishra P, Grover C, *et al*. A prospective study of cutaneous manifestations of spinal dysraphism from India. Pediatr Dermatol 2009;26:688-95.
13. Guggisberg D, Hadj-Rabia S, Viney C, Bodemer C, Brunelle F, Zerah M, *et al*. Skin markers of occult spinal dysraphism in children: A review of 54 cases. Arch Dermatol 2004;140:1109-15.
14. Vieira AR, Taucher SC. Maternal age and neural tube defects: Evidence for a greater effect on spina bifida than anencephly. Influence of maternal age on the risk for neural tube defects, a meta analysis. Rev Med Chile 2005;133:62-70.s